CATSPER2 gene

cation channel sperm associated 2

Normal Function

The *CATSPER2* gene provides instructions for producing a protein that is found in the tail (flagellum) of sperm cells. The CATSPER2 protein plays a role in sperm cell movement (motility) and is required for sperm cells to push through the outside membrane of the egg cell during fertilization. The CATSPER2 protein is embedded in the membrane of the sperm tail and is necessary in order for positively charged calcium atoms (calcium cations) to enter the cell. Calcium cations are needed for a type of sperm motility called hyperactivation. Hyperactivation is characterized by vigorous movements of the sperm tail, which are required for the sperm to push through the membrane of the egg cell during fertilization.

Health Conditions Related to Genetic Changes

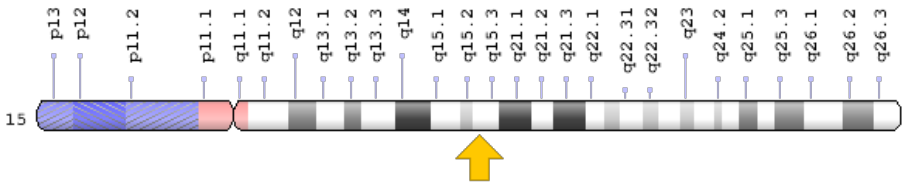
sensorineural deafness and male infertility

The symptoms of sensorineural deafness and male infertility are caused by a deletion of genetic material on the long (q) arm of chromosome 15. The chromosomal region that is typically deleted contains multiple genes, including the *CATSPER2* gene. People with this condition have the deletion in both copies of chromosome 15 in each cell. As a result of the deletion, affected individuals are missing both copies of the *CATSPER2* gene, and no CATSPER2 protein is produced. A lack of CATSPER2 protein impairs calcium entry into the sperm cell, which decreases motility and prevents hyperactivation. Lack of hyperactivation results in sperm that are unable to push through the membrane of the egg cell and achieve fertilization. These sperm abnormalities are the cause of infertility in affected males.

Chromosomal Location

Cytogenetic Location: 15q15.3, which is the long (q) arm of chromosome 15 at position 15.3

Molecular Location: base pairs 43,630,562 to 43,648,845 on chromosome 15 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cation channel sperm-associated protein 2
- cation channel, sperm associated 2
- CTSR2_HUMAN
- sperm ion channel

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Action at a Distance: Mammalian Gametes
  https://www.ncbi.nlm.nih.gov/books/NBK10010/?rendertype=box&id=A1381

GeneReviews

- CATSPER-Related Male Infertility
  https://www.ncbi.nlm.nih.gov/books/NBK22925

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CATSPER2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CATION CHANNEL, SPERM-ASSOCIATED, 2
  http://omim.org/entry/607249

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CATSPER2%5Bgene%5D
- HGNC Gene Family: Cation channels sperm associated
  http://www.genenames.org/cgi-bin/genefamilies/set/186
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18810
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/117155
- UniProt
  http://www.uniprot.org/uniprot/Q96P56

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14657366
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC299835/
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17098888
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2598039/

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/CATSPER2

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